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Myopathy presenting as developmental delay and short stature

K M Holt MBchB BSc *Clinical Medical Officer, Department of Community Child Health, Greenwich District Health Authority, Greenwich, UK*

Keywords: myopathy; developmental delay

Case presented to Section of Paediatrics, 27 November 1992

Introduction

Myopathic disease of the muscular dystrophy type was diagnosed in a 5-year-old girl presenting with motor difficulties in primary school. The case emphasizes the role of a strong community paediatric service in the detection of disability. It illustrates how developmental delay is not necessarily due to neglect or deprivation in children from socially disadvantaged families. Thorough paediatric assessment is essential in all children with developmental delay to exclude a physical cause.

Case report

Teachers were concerned about a 5-year-old girl in the reception class of a primary school. She had the following problems:

1 Dificulty in getting up from the ground.
2 Abnormal gait: she walked on tip toe with her arms extended.
3 Inability to run or dance.
4 She had a poor attention span and some learning difficulties.

The school doctor was asked to assess her. Past history had been that she tended to constipation: overflow soiling being a problem from the age of 2 years. The health visitor had been concerned about speech and motor delay. There was no cause found for these problems; but lack of stimulation at home and neglect had been thought to be underlying them. Since toddler age her motor skills had deteriorated; from being able to run to an inability to do so. Her gait has also changed; from normal to walking on tip toes.

A family history of muscular dystrophy was present: the maternal grandfather had a dystrophic disease rendering him wheelchair bound in his thirties.

Physical examination

A small, pale, fair-haired girl (height 3-10thC, weight=3rd centile) was noted to have difficulty in rising to stand. She was unable to hop.

She had proximal muscle weakness in her legs: with no focal neurological signs. She exhibited a partial Gowers manoeuvre (as described by Gower demonstrating the difficulty in rising from the floor).

She had bilateral tightening of her achilles tendons.

This clinical picture was suggestive of a muscular disease: reminiscent of a dystrophy.

Investigations were positive as follows:

1 Creatine kinase	20 500 (NR 25-175)
Repeated	2190 and 5043
2 Maternal creatine kinase	325

Needle muscle biopsy was abnormal, but not diagnostic of a specific disease. Abnormal features were: several foci of regenerating fibres; variation in fibre size; one cluster of histiocytes; polymyositis was excluded on the basis of the biopsy.

Deoxyribonucleic acid (DNA) analysis and dystrophin analysis was uninformative.

Discussion

An *X*-linked recessive condition was postulated in this girl due to the positive family history (affected maternal grandfather). The mother is clinically unaffected but has a mildly raised creatine kinase. Girls are rarely affected by *X*-linked conditions, but can be if they are 'manifest carriers' of the disease[1]. The commonest *X*-linked myopathic disorders with elevated creatine kinase and the clinical features that she presented are the muscular dystrophies Duchenne and Becker. Females can be manifest carriers of the Duchenne and Becker muscular dystrophies (DMD, BMD) if they carry an *X*-autosomal-translocation, where the breakpoint is at the DMD/BMD locus on the *X*-chromosome[2].

The clinical course of the grandfather's disease suggests BMD (clinical definition: patients who remain ambulant beyond 16 years)[3]. Further clarification is possible by DNA analysis using commercially available cDNA probes. Sixty-five per cent of BMDs have identifiable gene deletions of the dystrophy gene using these probes[5].

Assessment of the quality of dystrophin protein in muscle biopsy specimens is the definitive method for discrimination between DMB and BMD and other neuromuscular disorders[4,6]. BMD is due to a deficiency of dystrophin or alteration in its molecular mass. Further investigation needs to be done in this case to confirm the diagnosis.

Community paediatricians need to be trained both in general paediatrics and paediatric neurology. A strong community paediatric service is particularly important in areas of social deprivation where the school medical service acts as a safety net for children whose medical needs have not been met pre-school. Follow-up and management of such children may rely on the community paediatric service as access can sometimes only be obtained at school or at home. Building close links with the family can assist utilization of regional specialist centres for further investigation and management advice as is needed in this case.

Correspondence to Dr Kim M Holt, Senior Registrar in Paediatrics, Wythenshawe Health Care Centre, Stancliffe Road, Sharston, Manchester M22 4PR, UK

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Tuberculous fissure-*in ano*

S R Myers FRCS *The Department of General Surgery, The Norfolk and Norwich Hospital, Brunswick Road, Norwich NR1 3SR, UK*

Keywords: tuberculosis; gastrointestinal tuberculosis; perianal disease; anal ulcer

Summary

Anal tuberculosis is rare, with cases appearing sporadically in the worldwide literature, and only a single case report of the condition occurring in the UK over the last 10 years[1]. We are able to present a case of tuberculous fissure-*in ano*.

Case report

A 70-year-old male presented complaining of increasing pain and discharge from perianal eczema. He had been seen intermittently in a dermatology clinic over the previous 7 years with a diagnosis of perianal eczema, and treated with topical preparations including steroids. A widower, he was heterosexual, a non-smoker and consumed around 30 units of alcohol per week. He had no known contacts with tuberculosis.

On examination, he was well, apyrexial with no lymphadenopathy. Due to severe pain, an examination under anaesthetic was performed: sigmoidoscopy to 20 cm was normal, the perianal skin was thickened and hyperkeratotic, an oedematous hyperkeratotic skin tag was excised, a small posterior fissure was noted, and a large anterior fissure-*in ano* was punch-biopsied.

Serum investigations revealed a full blood count, liver and bone profile within normal limits, an erythrocyte sedimentation rate of 30 mm/h, and a C-reactive protein level of 4 mg/L. Human immunodeficiency virus (HIV) antibody testing was negative, and serum immunoglobulin levels normal. A chest X-ray had shown evidence of old calcified tuberculosis. Barium enema examination was unremarkable.

The histopathology report was of acanthotic, hyperkeratotic skin showing severe chronic inflammation composed, in part, of epithelioid cell and macrophage giant cell granulomata. Occasional organisms were stained by Ziehl-Nielson (Z-N) and modified Z-N techniques. There was no evidence of dysplasia or neoplasia.

Biopsy cultures were negative. A course of chemotherapy with isoniazid, rifampicin, pyrazinamide, and ethambutol was commenced, with resolution of symptoms within months but a persisting eczematous condition to date.

Correspondence to: Mr S R Myers FRCS, 124B Grove Park, Camberwell, London SE5, UK

Discussion

Reflecting the general decline in the incidence of tuberculosis, that of anal tuberculosis specifically had shown a sharp drop from a percentage incidence of tuberculous fistulae of 16% in 1921 to <1% in 1969[2]. However, non-European centres have reported figures as high as 17% in 1964[3] and 15.6% in 1988[4].

Anal tuberculosis has been subdivided into four categories by Bacon[5]: ulcerative, verrucous, lupoid, and miliary (the site, the virulence of the organism, and the number of bacilli possibly relating to the type of lesion that occurs[6]). Ulcerative tuberculosis is the commoner form, and is usually secondary to a focus in lung or intestine. Tuberculous fissure-*in ano* has been included in this group, but fistular disease has been categorized separately by some[1]. In the absence of a history of recent contact, and with radiological evidence of previous pulmonary tuberculosis, it seems likely that reactivation of tubercle originally ingested from the lung or spread by the haematogenous route was the mode of infection. HIV infection should be borne in mind[7].

Although fistular disease accounted for many of the early cases of anal tuberculosis in the West, it is now uncommon. However, recent experience in India demonstrates that fistulae there are relatively common, and that tuberculosis should be suspected in those with multiple fistulae, recurrent fistulae, concomitant pulmonary tuberculosis or inguinal lymphadenopathy[4].

The differential diagnosis includes: Crohn's disease, venereal lesions, neoplasia, foreign body reactions, and sarcoidosis.

Conclusion

In cases of intractable perianal disease, a high index of suspicion and adequate histopathological and microbiological assessment are required to exclude tuberculosis.

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